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Polymorphism of 11 non-CODIS STRs in a population sample of religious minority of Old Believers residing in northeastern Poland

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ABSTRACT

Purpose: The objective of this paper was to provide a database of 11 short tandem repeat (STR) markers not included in the Combined DNA Index System (non-CODIS) for a population sample of Old Believers (n=120) living in northeastern Poland.
Material and methods: Deoxyribonucleic acid (DNA) was extracted using Chelex-100 procedure. Genetic profiles were obtained using Mentype Chimera Kit (AG Biotype) and ABI 310 Genetic Analyzer. The statistical tests were performed using GDA v1.1 and PowerStats v1.2 software.
Results: The genotype frequency distributions showed no deviations from Hardy-Weinberg equilibrium (HWE) except for D5S2500 and D3S1744. The departures appeared statistically insignificant when the Bonferroni correction was used for the number of analysed loci. Significant differences between the Old Believers and Polish Caucasians were found at D7S1517, D8S1132, D2S1360, D5S2500, D6S474, D4S2366 and D3S1744. The combined values of matching probability (MP) and mean exclusion chance (MEC) are 8.35x10-15 and 0.999998, respectively.
Conclusions: A DNA database was established that may be used for the purpose of genetic profile comparison in criminal cases and chimerism monitoring after bone marrow transplantation. Significant differences revealed between the autochthonous Poles and the Old Believers by using RxC test and FST estimate support the idea of genetic isolation of this religious minority. Genetic polymorphisms analysed using statistical methods may be informative in differentiation of populations and ethnic groups in northeastern Poland.

Key words: forensic genetics, population genetics, STRs, Old Believers, northeastern Poland

INTRODUCTION

Old Believers are a group of Russian religious dissenters who refused to accept the liturgical reforms imposed upon the Russian Orthodox Church in 1653–66 by Nikon - the patriarch of Moscow. Old Believers became separated after 1666 from the hierarchy of the Russian Orthodox Church and split into a number of different sects living in close, restricted communities for centuries to avoid persecution [1]. Several groups have survived into modern times. Significant Old Believers communities exist in the US, Canada, Australia and South America. Smaller hidden communities have been found in northern Russia and Poland. Nowadays, nearly 600 Old Believers inhabit Suwalki region in northeastern Poland, where they founded several villages and have struggled to maintain their religious identity and traditional ways of life, such as farming and continuing to speak Russian [2].

Due to polymorphic nature of microsatellite markers (short tandem repeats, STRs) the percentage distributions of their observed alleles vary within and between populations and ethnic groups. In 1997, the Federal Bureau of Investigation (FBI) announced the selection of 13 STR loci to constitute the core of the United States national database called CODIS (Combined DNA Index System) [3]. The CODIS system has been widely adopted by forensic DNA analysts. The 13 CODIS loci used in the U.S. are CSF1PO, FGA, TH01, TPOX, VWA, D3S1358, D5S818, D7S820, D8S1179, D13S317, D16S539, D18S51 and D21S11 [4,5]. The U.K. and much of Europe

utilize 10 core loci that include the additional markers D2S1338 and D19S433 along with eight overlapping loci FGA, TH01, VWA, D3S1358, D8S1179, D16S539, D18S51 and D21S11 [5]. On the other hand, a number of new STR loci are under development to aid forensic casework and the human identity testing community. Laboratories worldwide are contributing to the analysis of STR allele frequencies in different human populations. Non-CODIS STR (NC) loci were chosen to be physically unlinked from the standard core CODIS STR loci so as to permit use of the product rule when combining data between CODIS STRs and NC loci.

In addition to forensic applications, genotyping of STRs has been applied as a standard procedure to determine cellular component ratio of donor and recipient in the management of patients receiving allogeneic bone marrow transplants [6,7]. Data analysis provides distinct STR genotypic profiles for the donor and for the transplant recipient. STR loci that are polymorphic (informative) between these individuals are used to assess relative amounts of recipient and donor DNA in the post-transplant sample. Full chimerism is used to refer to a patient which exhibits a post-transplant phenotype in hematopoietic cells that is all of donor origin. Mixed chimerism refers to a patient which exhibits a mixture of patient and donor phenotype in hematopoietic cells after transplant. The formula to calculate donor chimerism values is based on the different allelic distribution type between donor and recipient. With the commercial kits, many STR loci have informative alleles for both donor and recipient in the unrelated donor setting. As most hematopoietic stem cell transplantations (HSCTs) are performed between related donor transplant pairs, they have a higher tendency to inherit the same alleles at a specific locus. Thus, informativity in terms of chimerism analysis is usually substantially lower than that in forensic identification and is correlated with the degree of heterozygosity rather, than with the total number of alleles present [8,9]. These data indicate that selection of suitable STR markers is important to improve diagnostics based on STR analysis. The suitable forensic efficiency parameter - matching probability (MP) displays a coincidence that a random unrelated person would by chance have the same DNA profile as that obtained from the evidence, or else is the number of individuals that may be surveyed before finding the same DNA profile in a randomly encountered individual [10]. The combined MP for several loci is the product of the individual matching probability at each locus on the assumption that they are not linked.

The objective of this paper was to provide an 11 NC STR database for a population sample of Old Believers living in northeastern Poland for the purpose of genetic profile comparison in criminal cases and chimerism monitoring following HSCTs.

MATERIAL AND METHODS

Buccal swabs were collected from 120 unrelated healthy volunteers of the religious minority of Old Believers residing in northeastern Poland. DNA was extracted using the Chelex 100 and proteinase K protocol [11]. A commercially available kit Humantype Chimera (Biotype AG, Germany) developed as multiplex application for chimerism monitoring after bone marrow transplantation was used to co-amplify 12 STR loci: D2S1360, D3S1744, D4S2366, D5S2500, D6S474, D7S1517, D8S1132, D10S2325, D12S391, D18S51, D21S2055, SE33 (ACTBP8) and amelogenin. DNA templates (0.5-1.0ng) were amplified according to the manufacturer's instructions using PCR System 9700 (Applied Biosystems). Genotyping was performed in ABI 310 Genetic Analyzer (Applied Biosystems) using POP-4 and reference ladders. Genotyper v2.5 software was used with the macro included in the Humantype Chimera Template File. The biostatistical parameters were calculated using GDA v1.1 and Promega PowerStats v1.2 software [12,13]. The CODIS marker D18S51 was excluded from the analysis. For multiple comparisons, the original significance levels achieved (*P*-values) were transformed by the Bonferroni correction procedure [14]. Comparison of allele frequency distributions was performed by means of a pairwise population comparison test (RxC contingency test; G. Carmody, Ottawa, Canada).

RESULTS AND DISCUSSION

Observed allele frequencies of the 11 short tandem repeat loci and calculated values of the forensic efficiency parameters are displayed in *Tab. 1*. The genotype frequency distributions showed no deviations from (HWE) except for D5S2500 and D3S1744, based on the exact test (P=0.0450 and 0.0360, respectively). The departures appeared statistically insignificant when the Bonferroni correction was used for the number of analysed loci (i.e. 11 loci per database yields an actual significance level of 0.005). Pairwise comparison using the exact test disequilibrium analysis yielded departures from independence for pairs of loci D2S1360/D5S2500 and D3S1744/D6S474 (P=0.0060 and 0.0420, respectively), however neither of these values was significant after the Bonferroni adjustment. The inbreeding coefficient (F_{IS}) across the 11 markers was 0.048. Possible reasons could include sampling error, but considering that these loci showed an excess of homozygotes, the lack of equilibrium may be also due to inbreeding [15]. A pairwise testing for heterogeneity using the chi^2-test and the G-test revealed statistically significant differences between the Old Believers and Polish Caucasians for D7S1517, D8S1132, D2S1360, D5S2500, D6S474, D4S2366 and D3S1744 (P<0.05) [16,17,18]. Observed heterozygosity for all the systems ranged 0.688-0.942, with a mean value of 0.817, which is slightly lower than the average calculated for

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